In re Application of:

Peltonen et al.

Application No.: 09/509,595

Filed: July 5, 2000

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## In the Claims

Please amend claims 29 32, 34 38, 41-42, 44-46, 48, 54, and 62 as indicated in the following Listing of Claims.

PATENT

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Please cancel claims 39, 40, and 54, without prejudice or disclaimer.

The listing of claims will replace all prior version, and listings, of claims in the application:

## **Listing of Claims:**

- 29. (Currently Amended) A An isolated nucleic acid molecule encoding a polypeptide comprising two double paired zinc finger motifs, the nucleic acid molecule comprising or peptide thereof co-segregating in a mutated form with Autoimmune

  Polyendocrinopathy Candidiasis Ectodermal Dystrophy (APECED), which is selected from the group consisting of, comprising:
  - (a) a nucleic acid molecule comprising a nucleic acid molecule encoding the polypeptide having the amino acid sequence of SEQ ID NO:2;
  - (b) a nucleic acid molecule comprising a portion of the nucleic acid molecule having the nucleotide sequence of SEQ ID NO:1 that encodes the amino acid sequence of SEQ ID NO:2;
  - (b) a nucleic acid molecule hybridizing to the nucleic acid molecule of (a) or (b) in a hybridization solution comprising 50% formamide, 6X SSC, 0.1% SDS, and 100ug/ml single-stranded DNA and a wash solution comprising 0.1x SSC, 0.1% SDS wherein hybridization is performed at a temperature above 37C and washing is performed at a temperature above 55C; and
  - (c) a nucleic acid molecule which is degenerate to the nucleic acid molecule of (c).
- 30. (Currently Amended) The <u>isolated</u> nucleic acid molecule of claim 29, wherein said polypeptide has the function of a transcription factor or a transcription associated factor regulates or mediates transcription.

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- 31. (Currently Amended) The <u>isolated</u> nucleic acid molecule of claim 29 35, wherein said polypeptide comprises two double paired zinc finger motifs.
- 32. (Currently Amended) A An isolated nucleic acid molecule which is comprising the nucleotide sequence of a mammalian homolog of claim 29 SEQ ID NO:1, wherein the nucleic acid is complementary to a nucleic acid that hybridizes to SEQ ID NO:1 or SEQ ID NO:6 under low stringency conditions, and wherein the nucleic acid molecule is identical in sequence to a portion of human chromosome 21q22.3, or a portion of a mammalian chromosome that shares conserved synteny with human chromosome 21q22.3.
- 33. (Currently Amended) The <u>isolated</u> nucleic acid molecule of claim 32 wherein the molecule is a murine homologue.
- 34. (Currently Amended) The <u>isolated</u> nucleic acid molecule of claim 33 selected from the group consisting of:
  - (a) a nucleic acid molecule comprising a nucleic acid molecule encoding the polypeptide having the amino acid sequence of Fig. 14 SEQ ID NO:9;
  - (b) a nucleic acid molecule comprising the nucleic acid molecule having the nucleotide sequence of Fig. 14-SEQ ID NO:6 that encodes the amino acid sequence of Fig. 14; and
  - a nucleic acid molecule hybridizing to the nucleic acid molecule of (a) or (b) in a hybridization solution comprising 50% formamide, 6X SSC, 0.1% SDS, and 100ug/ml single-stranded DNA and a wash solution comprising 0.1x SSC, 0.1% SDS wherein hybridization is performed at a temperature above 37C and washing is performed at a temperature above 55C; and
  - (d) a nucleic acid molecule which is degenerate to the nucleic acid molecule of (c).

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- 35. (Currently Amended) A An isolated nucleic acid molecule deviating differing in nucleotide sequence by at least one nucleotide mutation from the nucleic acid molecule of elaim 29 SEQ ID NO:1, wherein said nucleic acid molecule hybridizes to SEQ ID NO:1 in a hybridization solution comprising 50% formamide, 6X SSC, 0.1% SDS, and 100ug/ml single-stranded DNA and a wash solution comprising 0.1x SSC, 0.1% SDS wherein hybridization is performed at a temperature above 37C and washing is performed at a temperature above 55C, wherein the nucleic acid molecule is identical in nucleotide sequence to a mutation mutated nucleic acid that co-segregates with APECED and is comprises
  - (i) an insertion;
  - (ii) a deletion;
  - (iii) a substitution; and/or or
  - (iv) an inversion

and wherein said mutation further results in a loss of function or a gain of function of the polypeptide encoded by a nucleic acid molecule of claim 29.

- 36. (Currently Amended) The <u>isolated</u> nucleic acid molecule of claim 35, wherein said insertion, which is a duplication of 4 nucleotides (CCTG) normally found at position 1086-1089, is a 4 nucleotide insertion at the nucleotide position 1085 or 1090, an insertion of an adenosine at position 1284, or an insertion of a cytosine at position 1365 of the nucleotide sequence of Fig. 2A SEQ ID NO:1.
- 37. (Currently Amended) The <u>isolated</u> nucleic acid molecule of claim 35, wherein said deletion is a 13 nucleotide deletion of nucleotides 1085-1097, a deletion of the thymidine at position 1051 or a deletion of the cytosine at position 1309 or 1313 of the nucleotide sequence of Fig. 2A SEQ ID NO:1.

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- 38. (Currently Amended) The <u>isolated</u> nucleic acid molecule of claim 35, wherein said substitution is a cytosine to thymidine exchange at nucleotide position 889, a guanosine to thymidine exchange at nucleotide position 358, an adenosine to guanosine exchange at nucleotide position 374, a guanosine to adenosine exchange at nucleotide position 1052, or a cytosine to adenosine exchange at nucleotide position 1094 of the nucleotide sequence of Fig. 2A SEQ ID NO:1.
- 39. (Cancel)
- 40. (Cancel)
- 41. (Currently Amended) A An isolated fragment of the nucleic acid molecule of claim 29 or claim 35 comprising at least about 14 21 nucleotides, wherein the isolated fragment hybridizes to SEQ ID NO:1 in a hybridization solution comprising 50% formamide, 6X SSC, 0.1% SDS, and 100ug/ml single-stranded DNA and a wash solution comprising 0.1x SSC, 0.1% SDS wherein hybridization is performed at a temperature above 37C and washing is performed at a temperature above 55C.
- 42. (Currently Amended) A An isolated nucleic acid molecule which is complementary to a nucleic acid molecule of claim 29 or claim 35.
- 43. (Currently Amended) The <u>isolated</u> nucleic acid molecule of claim 29 or claim 35 wherein the molecule is DNA or RNA.
- 44. (Currently Amended) A An isolated primer pair which hybridizes under stringent conditions to the nucleic acid molecule of any one of claims 29, 35, or 42

  SEQ ID NO:1 in a hybridization solution comprising 50% formamide, 6X SSC, 0.1%

  SDS, and 100ug/ml single-stranded DNA and a wash solution comprising 0.1x SSC,

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0.1% SDS wherein hybridization is performed at a temperature above 37C and washing is performed at a temperature above 55C.

- 45. (Currently Amended) A An isolated vector comprising the nucleic acid molecule of claim 29 or claim 35.
- 46. (Currently Amended) A An isolated host transformed with the a vector of claim 45.
- 47. (Previously Presented) The host of claim 46 which is a bacterium, a yeast cell, an insect cell, a fungal cell, a mammalian cell, a plant cell, a transgenic animal or a transgenic plant.
- 48. (Currently Amended) A method of producing a polypeptide of claim 29, or claim 35 comprising culturing the host of claim 46 an isolated host transformed with a vector comprising a nucleic acid molecule of claim 29 and isolating said polypeptide from said culture or said host.
- 49. (Withdrawn) A polypeptide produced by the method of claim 48.
- 50. (Withdrawn) A polypeptide encoded by the nucleic acid molecule of claim 29 or claim 35.
- 51. (Withdrawn) A compound derived from the polypeptide of claim 50 and having essentially the same three dimensional structure thereof.
- 52. (Withdrawn) An antibody that specifically recognizes the polypeptide of claim 50.
- 53. (Withdrawn) An antibody that specifically recognizes the compound of claim 51.

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- 54 (Cancel)
- 55. (Withdrawn) A method for testing for carriership for APECED or for a corresponding disease state comprising testing a sample obtained from a prospective patient or from a person suspected of carrying a predisposition for a mutation in the nucleic acid molecule of claim 29.
- 56. (Withdrawn) A method for testing for carriership for APECED or for a corresponding disease state comprising testing a sample obtained from a prospective patient or from a person suspected of carrying a predisposition for a mutated form of the polypeptide as defined in claim 29 in an immunoassay.
- 57. (Withdrawn) A pharmaceutical composition comprising the polypeptide of claim 50.
- 58. (Withdrawn) A pharmaceutical composition comprising the compound of claim 51.
- 59. (Withdrawn) A pharmaceutical composition comprising the antibody of claim 52.
- 60. (Withdrawn) The antibody of claim 52, wherein the antibody is monoclonal.
- 61. (Withdrawn) A method for treating a patient having APECED or being a carrier thereof comprising contacting a cell of the patient with a nucleic acid molecule of claim 29, thereby treating the patient.
- 62. (Currently Amended) An <u>isolated</u> nucleic acid molecule according to claim 29, wherein the nucleic acid molecule has the nucleotide sequence of SEQ ID NO:1.

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- 63. (New) An isolated nucleic acid molecule according to claim 29, wherein the encoded protein when expressed in COS cells is localized to punctate nuclear structures and vimentin filaments in the cytoplasm.
- 64. (New) An isolated nucleic acid molecule according to claim 29, wherein the nucleic acid molecule is identical in nucleotide sequence to a nucleic acid molecule that co-segregates in a mutated form with Autoimmune Polyendocrinopathy Candidiasis Ectodermal Dystrophy (APECED).
- 65. (New) An isolated nucleic acid molecule according to claim 32, wherein the isolated nucleic acid molecule is identical in nucleotide sequence to a portion of human chromosome 21q22.3.
- 66. (New) The nucleic acid molecule of claim 35, wherein said substitution is a cytosine to thymidine exchange at nucleotide position 889 of SEQ ID NO:1.
- 67. (New) An isolated nucleic acid molecule encoding a protein that mediates and regulates transcription, comprising two Cys4-His-Cys3 double paired finger motifs, wherein the encoded protein when expressed in COS cells is localized to punctate nuclear structures and vimentin filaments in the cytoplasm, and wherein the nucleic acid molecule is complementary to a nucleic acid molecule that hybridizes to SEQ ID NO:1 or SEQ ID NO:6 in a hybridization solution comprising 50% formamide, 6X SSC, 0.1% SDS, and 100ug/ml single-stranded DNA and a wash solution comprising 0.1x SSC, 0.1% SDS wherein hybridization is performed at a temperature above 37°C and washing is performed at a temperature above 55°C.